

THE UNIVERSITY OF ROCHESTER
SCHOOL OF MEDICINE AND DENTISTRY
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ROCHESTER, NEW YORK 14620

DEPARTMENT OF ANATOMY
DIVISION OF GENETICS

March 2, 1971

Victor A. McKusick, M.D.
Division of Medical Genetics
Department of Medicine
Johns Hopkins University
School of Medicine
Baltimore, Maryland 21205

Dear Vic:

In keeping with your memo requesting further suggestions for topics for the Fourth Annual Conference in the Clinical Delineation of Birth Defects, I would wonder whether you would be interested in a report of a rather unique family in which imperforate anus and bony anomalies of the hands and feet are dominantly inherited. In your provisional program I note that imperforate anus is being discussed and our family might well be of interest in that regard.

In essence, we have a family showing what may be called a Hand-Foot-Anus Syndrome. The father and four of his six children are all affected. Two of the affected children were prospectively ascertained in that they were born after we initially counseled the family in 1967.

We have not submitted a manuscript to any journal as yet, although Mr. Brocks and I did submit an abstract last year for the Society for Pediatric Research meeting. The abstract appeared, but by title only as it was not selected for the program.

I believe our family represents the first in which autosomal dominant inheritance of imperforate anus has been demonstrated in terms of an affected parent with affected children of both sexes. Moreover, the syndrome is quite different from that of Say and Gerald and is one which has not yet been reported.

As you know I am presently scheduled to present our findings on Trypsinogen Deficiency Disease and it remains my first choice of course. If the rules permit a second

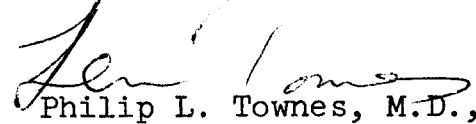
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paper, then I would request your consideration of Hand-Foot-Anus Syndrome by Philip L. Townes and Eric Brocks.

Thank you for your consideration.

Very best regards,

A handwritten signature in cursive script, appearing to read "Philip L. Townes".

Philip L. Townes, M.D., Ph.D.

PLT:ms